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Association of interleukin-6 gene polymorphism (*rs1800796*) with severity and functional status of osteoarthritis in elderly individuals

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Osteoarthritis (OA) is the most prevalent disease of the musculoskeletal system and it has an important genetic component. Despite several reports have shown the involvement of pro-inflammatory cytokine such as interleukin-1β and TNF-α, the role of interleukin-6 *(IL-6*) in osteoarthritis is still unclear. Thus, this study aimed to analyze the relationship between the single nucleotide polymorphism in the portion - 572 of the promoter region of the *IL-6* gene with hip and knee OA in the elderly. 257 physically independent elderly were recruited (92 individuals with osteoarthritis and 165 individuals without osteoarthritis). Blood samples were collected from patients for the DNA fragments extraction and amplification by qPCR. The degree of joint damage was assessed by radiographic classification based on the criteria of Kellgren and Lawrence. The functional status was evaluated by Lequesne and WOMAC questionnaires. It was observed that individuals carrying the C allele have lower susceptibility to osteoarthritis (OR = 0.51, 95% CI: 0.32- 0.80, p = 0.004) and less radiological impairment for both hip (Fisher-Freeman-Halton test = 4.2 and p=0.04) and knee joints (Fisher-Freeman-Halton test = 4.7 and p=0.03). Regarding functional status, individuals carrying the C allele has a lower degree of functional impairment assessed by WOMAC (p = 0.04). Additionally, it was observed a marked reduction in IL-6 serum levels in individuals with GC and CC genotypes when compared to individuals harboring GG genotype. In conclusion, the polymorphism -572G/C *IL-6* is a protective factor for the presence and severity of hip and knee osteoarthritis in the elderly.

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IRF5, PTPN22, CD28, IL2RA, KIF5A, BLK andTNFAIP3 genes polymorphisms and lupus susceptibility in a cohort from the Egypt delta; relation to other ethnic groups

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Objective: To replicate a single nucleotide polymorphism (SNP) of known genes for lupus (IRF5rs10488631, PTPN22rs2476601, BLKrs2736340 and TNFAIP3rs5029939) and other autoimmune diseases (CD28rs1980422, IL2RArs2104286 and KIF5Ars1678542) on a newly studied Egyptian cohort to investigate the genetic disparity with different studied ethnic groups in relation to lupus susceptibility.

Methods: 170 Egyptian patients from Egypt Delta with SLE and 241 matched healthy controls were genotyped by Taqman real time PCR for the selected SNPs.

Results: The results revealed significant association withIRF5 (p<0.0001) and PTPN22 (p=0.008) and insignificant association with KIF5A, CD28, IL2RA, BLK and TNFAIP3 genes.

Conclusions: This study may provide an additional evidence for the association between IRF5 and PTPN22 and lupus susceptibility and may exclude it for CD28, IL2RA and KIF5A.

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